

**Course Title:** Advanced Topics in Biomedical Ontology

Department of Biomedical Informatics  
Jacobs School of Medicine and Biomedical Sciences

**Course Subject Code:** BMI  
**Course Number:** 708  
**Type of Instruction:** SEM  
**Class Number:** 24641  
**Semester:** Fall 2021

**1** COURSE INFORMATION

- Date(s)/Time(s): September 1 – December 10, 2021 – Wednesdays 10am – 1pm
- Delivery Mode: Remote with recording
- Number of Credits: 3
- Course director / instructor: Werner Ceusters, MD (contact: [wceusters@gmail.com](mailto:wceusters@gmail.com))

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### 3 COURSE DESCRIPTION

- The course reviews applications and theories underlying biomedical knowledge representation and ontology. The methods and tools for applied ontology as well as the management and maintenance of biomedical ontologies will be discussed in detail thereby focusing on the principles of ontological realism and the implementation and axiomatization thereof in the Basic Formal Ontology (BFO2020).
- Students will gain hands-on implementation experience with the theories underlying the Information Artifact Ontology (IAO) and the Ontology for General Medical Science (OGMS). The Web Ontology Language (OWL) and the limitations thereof will be discussed and compared with implementations in Prolog able to query ontologies. The students will learn how to implement and evaluate classifiers and their role in subsumption. They will learn both the transitive and reflexive closure of subsumption and its applied use in ontology development, maintenance and use.
- This course provides to that end an in-depth review of current theories and research underlying the development of biomedical ontologies not based on ontological realism, in the first place SNOMED CT and the use thereof in clinical data repositories based on the Observational Medical Outcomes Partnership (OMOP) Common Data Model (CDM) including a critical analysis and evaluation thereof.
- Course prerequisites: BMI508 or PHI548 or PHI549.

### 4 STUDENT LEARNING OUTCOMES (SLO)

#### 4.1 *Course Learning Outcomes*

The following table lists the SLOs specifically for this course, thereby showing in which classes these SLOs will be covered and what degree of learning is aimed for (introduced, reinforced or mastered). The last column indicates the relationships of the SLOs with the assessment criteria for the assignments by means of which the SLOs will be assessed (see section 11 below).

Course Learning Outcome ID	Students will be able to:	Class (Introduced / Reinforced / Mastered)	Assessed
CLO1	Understand and apply the principals of Ontological Realism	• W4 (R) • W13 (M) • W5 (R)	A2:b.c
CLO2	Understand and apply the Basic Formal Ontology in ontology design	• W4 (R) • W12 (M) • W8 (R)	A2:b.c A3:b A4:b
CLO3	Understand and apply the Ontology of General Medical Science in biomedical ontology design	• W6 (R) • W12 (M)	A3:c.d A4:c.d
CLO4	Understand the strengths and weaknesses of prevailing biomedical data, information and knowledge management paradigms	• W1 (I) • W6 (R) • W2 (R) • W13 (M)	A1:a.c.d A3:e A5:a.b.c
CLO5	Identify opportunities, risks and challenges to current biomedical data, information and knowledge management paradigms	• W2 (I) • W6 (R) • W4 (R) • W12 (M) • W5 (R)	A2:c A4:f A5:a.b.c
CLO6	Assess the quality of existing clinical research data repositories using ontological principles	• W2 (I) • W12 (M) • W9 (R) • W13 (M) • W10 (R)	A3:f A4:f
CLO7	Develop ontology-based solutions for quality improvement of biomedical and clinical research data	• W3 (I) • W9 (R) • W7 (R) • W10 (M) • W8 (R)	A3:g A4:g A5:a.b.c
CLO8	Understand and apply the principles for life cycle management for biomedical ontologies	• W11 (M) • W12 (M)	A4:g

#### 4.2 *BMI PhD Program Outcomes / Competencies for the concentration in Biomedical Ontology*

The following table lists the SLOs for PhD students in Biomedical Informatics with a concentration in Biomedical Ontology. The table shows in which classes these SLOs will be covered and what degree of learning is aimed for (introduced, reinforced or mastered). The last column indicates the relationships of the SLOs with the assessment criteria for the assignments by means of which the SLOs will be assessed (see section 11 below).

<b>Outcome ID</b>	<b>BMI PhD Program Outcomes / Competencies In Biomedical Ontology</b>	<b>Class (Introduced / Reinforced / Mastered)</b>	<b>Assessed</b>
CSPO1	Methods of data representation, manipulation, storage, analysis and mining in healthcare and biomedical research databases	<ul style="list-style-type: none"> <li>• W1 (R)</li> <li>• W2 (R)</li> <li>• W3 (R)</li> <li>• W9 (R)</li> </ul> <ul style="list-style-type: none"> <li>• W10 (R)</li> <li>• W13 (M)</li> <li>• W14 (M)</li> </ul>	A1:a.c.d A3:e A4:e A5:a.b.c
CSPO2	Technical approaches to acquiring, modeling, representing and managing healthcare and biomedical research knowledge	<ul style="list-style-type: none"> <li>• W3 (I)</li> <li>• W7 (R)</li> </ul> <ul style="list-style-type: none"> <li>• W8 (R)</li> <li>• W12 (M)</li> </ul>	A3:f A4:f
CSPO3	Information retrieval and critical analysis skills	<ul style="list-style-type: none"> <li>• W2 (I)</li> </ul> <ul style="list-style-type: none"> <li>• W12 (R)</li> </ul>	A4:f.h
CSPO4	Ontological Realism, the Basic Formal Ontology (BFO), and the Ontology of General Medical Science	<ul style="list-style-type: none"> <li>• W4 (R)</li> <li>• W5 (R)</li> <li>• W6 (R)</li> <li>• W9 (R)</li> </ul> <ul style="list-style-type: none"> <li>• W10 (R)</li> <li>• W12 (M)</li> <li>• W14 (M)</li> </ul>	A2:b.c A3:b.c.d A4:b.c.d A5:a.b.c
CSPO5	Advanced methods and tools for managing biomedical ontologies (including the Web Ontology Language, OWL)	<ul style="list-style-type: none"> <li>• W4 (I)</li> </ul>	A2:b.c
CSPO6	Use of ontology editors and add-on tools (e.g., Protégé) to build a realism-based biomedical ontology	<ul style="list-style-type: none"> <li>• W3 (I)</li> <li>• W6 (R)</li> </ul> <ul style="list-style-type: none"> <li>• W7 (R)</li> <li>• W12 (M)</li> </ul>	A4:g
CSPO7	The principles for change management and upgrades to biomedical ontologies	<ul style="list-style-type: none"> <li>• W11 (M)</li> <li>• W12 (M)</li> </ul>	A4:g
CSPO8	The evaluation of biomedical ontologies and the published biomedical ontology research literature	<ul style="list-style-type: none"> <li>• W8 (R)</li> <li>• W14 (M)</li> </ul>	A5:a.b.c

#### 4.3 General BMI PhD program outcomes

The following table lists the SLOs for all PhD students in Biomedical Informatics, independent of concentration. The table shows in which classes these SLOs will be covered and what degree of learning is aimed for (introduced, reinforced or mastered). The last column indicates the relationships of the SLOs with the assessment criteria for the assignments by means of which the SLOs will be assessed (see section 11 below).

<b>ID</b>	<b>Description</b>	<b>Class (Introduced / Reinforced / Mastered)</b>	<b>Assessed</b>
PLO1	Have in-depth knowledge about and be able to discuss general key biomedical informatics concepts, models and theories and the major information management challenges and opportunities existing within various types of healthcare information systems	<ul style="list-style-type: none"> <li>• W1 (I)</li> <li>• W2 (R)</li> <li>• W6 (R)</li> </ul> <ul style="list-style-type: none"> <li>• W8 (R)</li> <li>• W9 (R)</li> <li>• W10 (R)</li> </ul>	A1:a.c.d A3:e.f A4:e.f
PLO2	Being able to apply advanced statistical data analysis and research methods to biomedical informatics problems in general and to the PhD student's core research domain in particular.	<ul style="list-style-type: none"> <li>• W10 (R)</li> <li>• W11 (R)</li> </ul>	
PLO3	The knowledge and skills needed to use information management systems and tools, and to implement effective information management systems within the scope of the biomedical informatics subspecialty the PhD students selected for their thesis.	<ul style="list-style-type: none"> <li>• W3 (I)</li> <li>• W4 (R)</li> <li>• W5 (R)</li> </ul> <ul style="list-style-type: none"> <li>• W7 (R)</li> <li>• W11 (R)</li> <li>• W12 (M)</li> </ul>	A2:b A4:e.f
PLO4	Master research project planning, management and completion in Biomedical Informatics.	<ul style="list-style-type: none"> <li>• Not applicable</li> </ul>	
PLO5	Advanced understanding of cutting-edge techniques and technologies to address difficult problems pertaining to the biomedical informatics subspecialty the PhD students selected for their thesis.	<ul style="list-style-type: none"> <li>• W3 (I)</li> <li>• W6 (R)</li> <li>• W8 (R)</li> <li>• W9 (R)</li> </ul> <ul style="list-style-type: none"> <li>• W12 (R)</li> <li>• W13 (M)</li> <li>• W14 (M)</li> </ul>	A3:f.g A4:f.g A5:a.b.c
PLO6	Ability to complete the PhD program successfully.	<ul style="list-style-type: none"> <li>• Not applicable</li> </ul>	

#### 4.4 Institutional learning outcomes

The following table outlines the Institutional Outcomes set forth for graduate students at UB. The table shows in which classes these SLOs, where applicable, will be covered and what degree of learning is aimed for (introduced, reinforced or mastered). The last column indicates the relationships of the SLOs with the assessment criteria for the assignments by means of which the SLOs will be assessed (see section 11 below).

ID	Learning outcome	Class (Introduced / Reinforced / Mastered)	Assessed
ILO1	<b>Critical Reasoning</b> — Demonstrate domain expertise, including critical reasoning and analysis.	<ul style="list-style-type: none"> <li>• W4 (M)</li> <li>• W5 (M)</li> <li>• W9 (M)</li> </ul> <ul style="list-style-type: none"> <li>• W10 (M)</li> <li>• W13 (M)</li> <li>• W14 (M)</li> </ul>	A1:a.b.c.d A2:d A3:h A4:h A5:a.b.c
ILO2	<b>Literacy Skills</b> — Apply effective communication, information, and digital literacy skills.	<ul style="list-style-type: none"> <li>• W4 (M)</li> <li>• W5 (M)</li> </ul> <ul style="list-style-type: none"> <li>• W14 (M)</li> </ul>	A1:b A2:d A3:h A4:h A5:a.b.c
ILO3	<b>Ethics and Responsibility</b> — Demonstrate ethical and professional responsibility and act according to the norms of the chosen discipline.	<ul style="list-style-type: none"> <li>• Not applicable</li> </ul>	A1:a A2:a A3:a A4:a
ILO4	<b>Local and Global Diversity</b> — Recognize the relevance of human and cultural diversity within local and global contexts.	<ul style="list-style-type: none"> <li>• Not applicable</li> </ul>	
ILO5	<b>Collaborate Positively</b> — Collaborate positively with others to achieve a common purpose.	<ul style="list-style-type: none"> <li>• W9 (M)</li> <li>• W14 (M)</li> </ul>	A3:i A5:a.b.c
ILO6	<b>Personal Skills</b> — Assess, articulate, and acknowledge personal skills, abilities and growth areas.	<ul style="list-style-type: none"> <li>• Not applicable</li> </ul>	
ILO7	<b>Service Engagement</b> — Demonstrate commitment to community service and engagement	<ul style="list-style-type: none"> <li>• Not applicable</li> </ul>	

## 5 COURSE REQUIREMENTS

- Students are required to read 10 papers and electronic publications as listed in the course materials in section 12 below.
- Students must attend all classes and must participate in class discussions. See attendance policy regulations for exceptions in section 13 below.
- Some classes may include a surprise in-class test, i.e. a test during the scheduled class time or a short assignment to be done on the spot. Students can earn extra credits for these, but will not be penalized in case of partial or total failure.
- All assignments need to be completed prior to the deadline specified in the course schedule and send electronically to [wceusters@gmail.com](mailto:wceusters@gmail.com). Thus no Google doc links or any other link to a cloud server.
- The filename of assignments should be formatted in the following way: BMI708-[number of the assignment]-[your UBIT name].[file-extension].  
For example, if the course director were a student and the requested file a Word document: “BMI708-A1-ceusters.docx”.
- The first line in the documents should always contain your full name (first and last name) and email address.
- Some assignments will consist of implementations in SWI-Prolog (<https://www.swi-prolog.org/Download.html>). They need to be completed by means of a coding editor of your choice. If you don't have one, I recommend for maximal functionality NotePad++ (<https://notepad-plus-plus.org/downloads/>). Implementation files must have the extension ‘.pl’ and follow the filename guidance stated above. They must be load-error-free, i.e., clicking on the filename when received should cause no warning or error from the Prolog interpreter.
- Since all assignments require documentation and rationales for design choices made, it is advised that the ‘.pl’ file contains short coding comments, that the entire implementation file is copied in a Word document, and that extensive documentation is then provided in that Word document. Both files, each with the same name except for the extension, should then be submitted at the same time, in the same email.
- Since most assignments build further on previous ones, good documentation of your work will help you in picking up where you left.
- The ‘sent’ date in the metadata header of the message through which the completed assignments will be received will be taken for assessment of in-time delivery.
- When assignments are delivered past the due date, a penalty of 1% of the positive final score will be applied for every 24 hours of delay. When such assignment is not delivered at all, the penalty will be equal to the % for which the assignment counts towards the final grade. Students unable to meet the deadline must inform the instructor by email and provide a valid reason prior to reaching the deadline. An alternative assignment and/or due date can then be agreed upon, be it entirely to the discretion of the instructor.
- You may email me at any time, exclusively at [wceusters@gmail.com](mailto:wceusters@gmail.com). Mails related to the class should in the subject line be prefixed with ‘BMI708:’, otherwise they will be ignored and not answered.
- The course will primarily consist of short lectures and much longer guided exercises. These exercises, together with the assignments A1 to A4, are selected in such a way that they contribute content-wise to the final assignment A5 to a large

extent. Students should keep this evolutionary aspect in mind and are therefore advised to document their assignments and corrections thereof together with material offered during class in such a way that their individual section of A5 (see section 11.5 below) is almost completed by the end of W12 so that they then can focus on the collaborative parts.

## **6 GRADING POLICY**

Grading follows standard graduate policies (<http://grad.buffalo.edu/Academics/Policies-Procedures/Grading-Procedures.html>). The following break-down will be used:

Final score weighting	Assignment
3%	A1
3%	A2
3%	A3
3%	A4
88%	A5
100%	

Extra credits obtained will be added after all assignments have been completed and assessed. Final scores may be curved upwards by the instructor upon his discretion.

Final Grades:

Grade	Quality Points	Percentage
A	4.0	93.0% -100.00%
A-	3.67	90.0% - 92.9%
B+	3.33	87.0% - 89.9%
B	3.00	83.0% - 86.9%
B-	2.67	80.0% - 82.9%
C+	2.33	77.0% - 79.9%
C	2.00	73.0% - 76.9%
C-	1.67	70.0% - 72.9%
D+	1.33	67.0% - 69.9%
D	1.00	60.0% - 66.9%
F	0	59.9% or below

An interim grade of Incomplete (I) may be assigned if the student has not completed all requirements for the course. An interim grade of 'I' shall not be assigned to a student who did not attend the course. The default grade accompanying an interim grade of 'I' shall be 'U' and will be displayed on the UB record as 'IU.' The default Unsatisfactory (U) grade shall become the permanent course grade of record if the 'IU' is not changed through formal notice by the instructor upon the student's completion of the course.

Assignment of an interim 'IU' is at the discretion of the instructor. A grade of 'IU' can be assigned only if successful completion of unfulfilled course requirements can result in a final grade better than the default 'U' grade. The student should have a passing average in the requirements already completed. The instructor shall provide the student specification, in writing, of the requirements to be fulfilled.

## **7 ACADEMIC INTEGRITY**

Academic integrity is a fundamental university value. Through the honest completion of academic work, students sustain the integrity of the university while facilitating the university's imperative for the transmission of knowledge and culture based upon the generation of new and innovative ideas. See <http://grad.buffalo.edu/Academics/Policies-Procedures/Academic-Integrity.html>.

Students may collaborate for the assignments in which case the submitted materials should be clearly labeled as such, with the names of all collaborating students. In case students who collaborate cannot come to a consensus for certain parts of the work, alternate solutions proposed by individual students should be clearly marked as such. Grading of individual students will take into account such alternatives.

## **8 ACCESSIBILITY RESOURCES**

If you have any disability which requires reasonable accommodations to enable you to participate in this course, please contact the Office of Accessibility Resources, 25 Capen Hall, 645-2608, and also the instructor of this course. The office will provide you with information and review appropriate arrangements for reasonable accommodations.

## 9 COURSE FEES

Standard UB tuition and fees. No extra costs.

## 10 COURSE ORGANIZATION / SCHEDULE

Reference: <http://registrar.buffalo.edu/calendars/academic/>

Week	Covered SLOs (Level)	Topics	Pre-class requirements	Educational method	Assignment
W1 Sep 1	CLO4 (I) CSPO1 (R) PLO1 (I)	<ul style="list-style-type: none"> <li>House keeping</li> <li>Course overview</li> <li>Structure and content of final evaluation report (A5) and how to get there.</li> <li>Structure of SNOMED-CT</li> </ul>	<ul style="list-style-type: none"> <li>Familiarize yourself with, and have available during class: <ul style="list-style-type: none"> <li><a href="https://browser.ihtsdotools.org/">https://browser.ihtsdotools.org/</a></li> <li><a href="https://confluence.ihtsdotools.org/display/DOCEG/SNOMED+CT+Editorial+Guide">https://confluence.ihtsdotools.org/display/DOCEG/SNOMED+CT+Editorial+Guide</a></li> </ul> </li> </ul>	<ul style="list-style-type: none"> <li>Lecture</li> <li>Guided exercises</li> </ul>	A1
W2 Sep 8	CLO4 (R) CLO5 (I) CLO6 (I) CSPO1 (R) CSPO3 (I) PLO1 (R)	<ul style="list-style-type: none"> <li>Annotating data repositories</li> </ul>	<ul style="list-style-type: none"> <li>Read R1 (<a href="https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6307753/">https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6307753/</a>)</li> </ul>	<ul style="list-style-type: none"> <li>Discussion of A1</li> <li>Lecture on problems with prevailing biomedical data representations</li> </ul>	
W3 Sep 15	CLO7 (I) CSPO1 (R) CSPO2 (I) CSPO6 (I) PLO3 (I) PLO5 (I) ILO1 (M)	<ul style="list-style-type: none"> <li>Gentle introduction to Common Logic and Prolog.</li> <li>Implementation of BFO2020's basic assumptions in Prolog.</li> </ul>	<ul style="list-style-type: none"> <li>Install SWI-Prolog (<a href="https://www.swi-prolog.org/Download.html">https://www.swi-prolog.org/Download.html</a>)</li> <li>Download BFO2020 (<a href="https://github.com/BFO-ontology/BFO-2020/archive/refs/heads/master.zip">https://github.com/BFO-ontology/BFO-2020/archive/refs/heads/master.zip</a>)</li> </ul>	<ul style="list-style-type: none"> <li>Lecture</li> <li>Guided hands-on exercises using BFO2020's 'existence-instantiation.cl' in '.../master.zip'</li> </ul>	
W4 Sep 22	CLO1 (R) CLO2 (R) CLO5 (R) CSPO4 (R) CSPO5 (I) PLO3 (R) ILO1 (M) ILO2 (M)	<ul style="list-style-type: none"> <li>Comparing SNOMED CT's, ICD's and BFO2020's basic assumptions.</li> <li>Introduction to creating knowledge source-compatible logical definitions, axioms, mappings and reasoning strategies in Prolog.</li> </ul>	<ul style="list-style-type: none"> <li>Read R2, R3 and R4</li> <li>(<a href="https://pubmed.ncbi.nlm.nih.gov/25160346/">https://pubmed.ncbi.nlm.nih.gov/25160346/</a>)</li> <li>(<a href="https://www.researchgate.net/publication/220438613_Consolidating_SNOMED_CT's_ontological_commitment">https://www.researchgate.net/publication/220438613_Consolidating_SNOMED_CT's_ontological_commitment</a>)</li> <li>(<a href="https://www.nature.com/articles/npre.2009.3465.1">https://www.nature.com/articles/npre.2009.3465.1</a>)</li> </ul>	<ul style="list-style-type: none"> <li>Lecture</li> <li>Guided hands-on exercises</li> </ul>	A2
W5 Sep 29	CLO1 (R) CLO5 (R) CSPO4 (R) PLO3 (R) ILO1 (M) ILO2 (M)	<ul style="list-style-type: none"> <li>Advanced examples in creating knowledge source-compatible logical definitions, axioms, mappings and reasoning strategies in Prolog</li> </ul>		<ul style="list-style-type: none"> <li>Discussion of A2</li> <li>Hands-on exercises</li> </ul>	
W6 Oct 6	CLO3 (R) CLO4 (R) CLO5 (R) CSPO4 (R) CSPO6 (R) PLO1 (R) PLO5 (R)	<ul style="list-style-type: none"> <li>SNOMED CT's disease (disorder) hierarchy compared with The Ontology of General Medical Science (OGMS)</li> </ul>	<ul style="list-style-type: none"> <li>Read R5 and R6: <ul style="list-style-type: none"> <li>(<a href="https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3041577/">https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3041577/</a>)</li> <li>(<a href="https://pubmed.ncbi.nlm.nih.gov/30170591/">https://pubmed.ncbi.nlm.nih.gov/30170591/</a>)</li> </ul> </li> </ul>	<ul style="list-style-type: none"> <li>Guided discussion on mistakes in R6</li> <li>Exercises in adequate mapping</li> </ul>	
W7 Oct 13	CLO7 (R) CSPO2 (R) CSPO6 (R) PLO3 (R)	<ul style="list-style-type: none"> <li>Implementing an adequate SNOMED CT – OGMS mapping</li> </ul>		<ul style="list-style-type: none"> <li>Guided exercise</li> </ul>	
W8 Oct 20	CLO2 (R) CLO7 (R) CSPO2 (R) CSPO8 (R) PLO1 (R) PLO5 (R)	<ul style="list-style-type: none"> <li>SNOMED CT and the principles of the Information Artifact Ontology.</li> </ul>	<ul style="list-style-type: none"> <li>Read R7 and R8</li> <li>(<a href="https://hal.archives-ouvertes.fr/hal-03041527/document">https://hal.archives-ouvertes.fr/hal-03041527/document</a>)</li> <li>(<a href="http://ceur-ws.org/Vol-1515/regular10.pdf">http://ceur-ws.org/Vol-1515/regular10.pdf</a>) (video of presentation: <a href="https://www.youtube.com/watch?v=PBKsupBquok">https://www.youtube.com/watch?v=PBKsupBquok</a>)</li> </ul>	<ul style="list-style-type: none"> <li>Lecture</li> <li>Guided implementation exercise</li> </ul>	
W9 Oct 27	CLO6 (R) PLO1 (R) CLO7 (R) PLO5 (R) CSPO1 (R) ILO1 (M) CSPO4 (R) ILO5 (M)	<ul style="list-style-type: none"> <li>Representing diagnoses and mis-diagnoses</li> </ul>	<ul style="list-style-type: none"> <li>Read R9 (<a href="https://jbiomedsem.biomedcentral.com/articles/10.1186/s13326-016-0098-5">https://jbiomedsem.biomedcentral.com/articles/10.1186/s13326-016-0098-5</a>)</li> </ul>	<ul style="list-style-type: none"> <li>Guided exercise</li> </ul>	A3

W10 Nov 3	CLO6 (R) CLO7 (M) CSPO1 (R) CSPO4 (R)	PLO1 (R) PLO2 (R) ILO1 (M)	<ul style="list-style-type: none"> <li>OMOP's 'condition status' diagnoses</li> </ul>		<ul style="list-style-type: none"> <li>Discussion of A3</li> </ul>	
W11 Nov 10	CLO8 (M) CSPO7 (M) PLO1 (R) PLO2 (R) PLO3 (R)		<ul style="list-style-type: none"> <li>Implementing principles for change management in ontologies.</li> </ul>	<ul style="list-style-type: none"> <li>Read R10 (<a href="https://www.nature.com/articles/41598-018-23395-2">https://www.nature.com/articles/41598-018-23395-2</a>)</li> </ul>	<ul style="list-style-type: none"> <li>Discussion of R10</li> <li>Implementation exercises</li> </ul>	
W12 Nov 17	CLO2 (M) CLO3 (M) CLO5 (M) CLO6 (M) CLO8 (M) CSPO2 (M)	CSPO3 (R) CSPO4 (M) CSPO6 (M) CSPO7 (M) PLO3 (M) PLO5 (R)	<ul style="list-style-type: none"> <li>Application ontology for mapping OMOP CDM6's clinical data tables to SNOMED CT, IAO and OGMS.</li> </ul>	<ul style="list-style-type: none"> <li>Study <a href="https://ohdsi.github.io/CommonDataModel/cdm60.html#Clinical_Data_Tables">https://ohdsi.github.io/CommonDataModel/cdm60.html#Clinical_Data_Tables</a></li> </ul>	<ul style="list-style-type: none"> <li>Guided exercise</li> </ul>	A4
W13 Dec 1	CLO1 (M) CLO4 (M) CLO6 (M) CSPO1 (M) PLO5 (M) ILO1 (M)		<ul style="list-style-type: none"> <li>Using ontological principles to determine the quality of clinical research data repositories based on OMOP's CDM6.</li> </ul>		<ul style="list-style-type: none"> <li>Discussion of the A4 assignments</li> </ul>	
W14 Dec 8	CSPO1 (M) CSPO4 (M) CSPO8 (M) PLO5 (M)	CLO4 (M) ILO1 (M) ILO2 (M) ILO5 (M)	<ul style="list-style-type: none"> <li>Course wrap-up</li> <li>Review of structure and content of A5</li> <li>Course evaluation by students</li> </ul>		<ul style="list-style-type: none"> <li>Discussion</li> </ul>	A5

## II ASSIGNMENTS

### 11.1 A1: Text annotation exercise using pre-coordinated SNOMED CT codes

- Annotate your designated case-reports (selected by the instructor according to your research interests) by using in a first step only pre-coordinated SNOMED CT codes such that at least 80% of the SNOMED CT semantic tags are covered by your annotations (see <https://confluence.ihtsdotools.org/display/DOCEG/Semantic+Tag>), and by linking in a second step, where applicable, codes derived from the first step by means of SNOMED CT associative relationships. Response format will be discussed in class. Assignment to be done strictly individually.
- Due date: Sept 6, 2021, noon.
- Assessment:

Assessment criteria	Weight	Assessed SLOs
a. Coverage of the semantic tag variety	(x%)	CLO4 PLO1 CSPO1 ILO1 ILO3
b. Rationales for (1) in-text term or phrase selection, (2) selected SNOMED CT code(s) for the corresponding in-text term, and (3) selection of code/associative relation/code triples.	x*50%	ILO1 ILO2
c. Correctness of selected SNOMED CT code(s) for the corresponding in-text term and the proposed associative relation triples.	x*20%	CLO4 PLO1 CSPO1 ILO1
d. Degree of specificity of the selected codes.	x*30%	CLO4 PLO1 CSPO1 ILO1

### 11.2 A2. Understanding SNOMED CT's Expression Constraint Language

- Assignment to be done strictly individually. Implement in Prolog the named simple expression constraints except 'self', 'any' and 'member of', thus eight in total, of SNOMED CT's Expression Constraint Language (ECL) listed on <https://confluence.ihtsdotools.org/pages/viewpage.action?pageId=110341487>. Apply this to the BFO2020 continuant hierarchy. Response format will be discussed in class.
- Due date: Sept 27, 2021, noon
- Assessment:

Assessment criteria	Weight	Assessed SLOs
a. Coverage of the eight simple named expression constraints	(x%)	ILO3
b. Correctness of the Prolog representation of the BFO2020 continuant hierarchy in line with the relevant formal definitions and axioms	30%	CLO1 CSPO4 CLO2 CSPO5 PLO3

c. Correctness of the implementation of the expression constraints with respect to the relevant BFO2020 assumptions, formal definitions and axioms	x*50%	CLO1 CLO2 CLO5	CSPO4 CSPO5 PLO3
d. Convincing arguments that the implementation is correct	x*20%	ILO1	ILO2

### 11.3 A3. Diagnosis types in the OMOP CDM

- Group assignment: implement in Prolog the formal definitions for the 9 OMOP concepts displayed at <https://athena.ohdsi.org/search-terms/terms?domain=Condition+Status&page=1&pageSize=15&query=diagnosis> that do not start with 'primary' or 'secondary', thereby ensuring that the perspectives provided by IAO, OGMS, SNOMED CT and OMOP are all fully preserved and mapped. Beware of several pitfalls and cans of worms!
- Due date: Nov 1, 2021, noon
- Assessment:

Assessment criteria	Weight	Assessed SLOs	
a. Coverage of the required 9 types	(x%)	ILO3	
b. Adherence of the implementation to BFO	20%	CLO2	CSPO4
c. Adherence of the implementation to IAO	10%	CLO3	CSPO4
d. Adherence of the implementation to OGMS	20%	CLO3	CSPO4
e. Coverage of the relevant SNOMED CT diagnostic types	10%	CLO4 PLO1	CSPO1
f. Correctness of the mapping of all sources	x*20%	CLO6 PLO1	CSPO2 PLO5
g. Correctness of the implementation	x*10%	CLO7	PLO5
h. Convincing arguments that the mapping is correct	x*10%	ILO1	ILO2
i. Total score		ILO5	

### 11.4 A4. Mapping OMOP information model to realism-based ontologies

- Complete and use the mapping developed in W12. Represent the by the instructor delineated part of the designated case report from A1 in Prolog so that the case can be queried through the mapping for all 4 perspectives (IAO, OGMS, SNOMED CT and OMOP). Make sure that any necessary changes to the ontology developed in previous assignments is represented in line with the ontology change management guidelines discussed in class. Assignment to be done strictly individually.
- Due date: Dec 1, 2021, noon.
- Assessment:

Assessment criteria	Weight	Assessed SLOs	
a. Coverage of the designated part and relevant SNOMED CT types and OMOP clinical table components	(x%)	ILO3	
b. Adherence of the implementation to BFO	20%	CLO2	CSPO4
c. Adherence of the implementation to IAO	10%	CLO3	CSPO4
d. Adherence of the implementation to OGMS	20%	CLO3	CSPO4
e. Coverage of the mapping of all sources	10%	CLO5 PLO1	PLO3 CSPO1
f. Correctness of the mapping of all sources	x*20%	CLO5 CLO6 PLO1 PLO3	CSPO2 CSPO3 PLO5
g. Correctness of the implementation, including adherence to ontology life cycle management	x*10%	CLO7 CLO8	PLO5 CSPO6 CSPO7
h. Convincing arguments that the mapping is correct	x*10%	CSPO3 ILO1	ILO2

### 11.5 A5. Evaluation report

- Group assignment: evaluation report on the BFO, OGMS, IAI, SNOMED CT and OMOP integration through ontology, describing all aspects covered in the course, following the structure described in W1. Building further on A4, students will describe their individual results – one per student – related to the covered case reports as if they were part of a 'preliminary result' section in a grant proposal including a discussion about alternatives, even when wrong, considered during the course. Students may work together on these individual sections so as to identify more issues and/or provide better rationales, but will be



separately assessed on the final content. They will then in an additional section compare the solutions and discuss pro's and con's. They will finally discuss ways for harmonization of the representations. This report will qualify as final exam.

- Due date: December 20, 2021, noon.
- Only ONE report is to be submitted, co-authored by ALL students. The filename should omit the UBIT-name part.
- Assessment:

Assessment criteria	Weight	Assessed SLOs
a. Clarity and faithfulness of the individual sections (separate score per student)	40%	CSPO8 CLO4 CLO5 CLO7
b. Completeness of the pro's and con's section with adequate argumentation (same score for all students)	30%	ILO1 CSPO1 ILO2 CSPO8
c. Soundness of the proposed harmonization (same score for all students)	30%	ILO5 PLO5 CSPO4

## 12 COURSE MATERIALS

This course requires the following books, papers and electronic publications, all of which are available publicly or through the UB Libraries:

- R1. Miñarro-Giménez JA, Martínez-Costa C, Karlsson D, Schulz S, Gøeg KR. Qualitative analysis of manual annotations of clinical text with SNOMED CT. PLoS One. 2018 Dec 27;13(12):e0209547. doi: 10.1371/journal.pone.0209547. PMID: 30589855; PMCID: PMC6307753.  
(<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6307753/>)
- R2. Schulz S, Rodrigues JM, Rector A, Spackman K, Campbell J, Ustün B, Chute CG, Solbrig H, Della Mea V, Millar J, Brand Persson K. What's in a class? Lessons learnt from the ICD - SNOMED CT harmonisation. Stud Health Technol Inform. 2014;205:1038-42. PMID: 25160346.  
(<https://pubmed.ncbi.nlm.nih.gov/25160346/>)
- R3. Schulz, Stefan, Cornet, Ronald, and Spackman, Kent. 'Consolidating SNOMED CT's Ontological Commitment'. 1 Jan. 2011 : 1 – 11.  
([https://www.researchgate.net/publication/220438613\\_Consolidating\\_SNOMED\\_CT's\\_ontological\\_commitment](https://www.researchgate.net/publication/220438613_Consolidating_SNOMED_CT's_ontological_commitment))
- R4. Schulz, S., Cornet, R. SNOMED CT's Ontological Commitment. Nat Prec (2009).  
(<https://www.nature.com/articles/npre.2009.3465.1>).
- R5. Scheuermann RH, Ceusters W, Smith B. Toward an ontological treatment of disease and diagnosis. Summit Transl Bioinform. 2009 Mar 1;2009:116-20.  
(<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3041577/>)
- R6. El-Sappagh S, Franda F, Ali F, Kwak KS. SNOMED CT standard ontology based on the ontology for general medical science. BMC Med Inform Decis Mak. 2018 Aug 31;18(1):76. doi: 10.1186/s12911-018-0651-5. PMID: 30170591; PMCID: PMC6119323.  
(<https://pubmed.ncbi.nlm.nih.gov/30170591/>)
- R7. Fumiaki Toyoshima, Adrien Barton, Olivier Grenier. Foundations for an Ontology of Belief, Desire and Intention. 11th International Conference on Formal Ontology in Information Systems (FOIS 2020), Sep 2021, Bolzano, Italy. pp.140-154, ff10.3233/faia200667ff. fhal-03041527f.  
(<https://hal.archives-ouvertes.fr/hal-03041527/document>)
- R8. Smith B, Ceusters W. Aboutness: Towards Foundations for the Information Artifact Ontology. International Conference on Biomedical Ontologies, ICBO 2015, Lisbon, Portugal, July 27-30, 2015;47-51  
(<http://ceur-ws.org/Vol-1515/regular10.pdf>)
- R9. Hogan WR and Ceusters W. Diagnosis, misdiagnosis, lucky guess, hearsay, and more: an ontological analysis. Journal of Biomedical Semantics 2016;7(54).  
(<https://jbiomedsem.biomedcentral.com/articles/10.1186/s13326-016-0098-5>)
- R10. Tomczak, A., Mortensen, J.M., Winnenburg, R. et al. Interpretation of biological experiments changes with evolution of the Gene Ontology and its annotations. Sci Rep 8, 5115 (2018). <https://doi.org/10.1038/s41598-018-23395-2>.  
(<https://www.nature.com/articles/s41598-018-23395-2>)

## 13 ATTENDANCE POLICY

Students are expected to attend *all* lectures and exercises. For religious observances, university sanctioned events, athletic commitments and family/work obligations/emergencies, absences may be granted upon request but can have an effect on the finally obtained grade (see grading policy)

For course cancellation/emergency planning, see the university website for cancellations/delays due to weather or other unforeseen events (<http://emergency.buffalo.edu/campus-weather-alerts.html>)